

## List of Publications by Year in descending order

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62  
papers

2,006  
citations

304743

22  
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254184

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63  
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docs citations

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times ranked

3344  
citing authors

#	ARTICLE	IF	CITATIONS
1	Early-onset leukoencephalomyelopathy due to a biallelic <i>NDUFV1</i> variant in a mid-forties patient. <i>Annals of Clinical and Translational Neurology</i> , 2022, 9, 888-892.	3.7	1
2	Erleichterung und Sorge, Hoffnung und Angst – der Weg zur Diagnosestellung einer SLC16A2-Mutation. <i>Paediatrica</i> , 2021, 32, .	0.1	0
3	Absence of Genotype/Phenotype Correlations Requires Molecular Diagnostic to Ascertain Stargardt and Stargardt-Like Swiss Patients. <i>Genes</i> , 2021, 12, 812.	2.4	0
4	Triheptanoin – Novel therapeutic approach for the ultra-rare disease mitochondrial malate dehydrogenase deficiency. <i>Molecular Genetics and Metabolism Reports</i> , 2021, 29, 100814.	1.1	5
5	A Novel Van der Woude Syndrome-Causing IRF6 Variant Is Subject to Incomplete Non-sense-Mediated mRNA Decay Affecting the Phenotype of Keratinocytes. <i>Frontiers in Cell and Developmental Biology</i> , 2020, 8, 583115.	3.7	7
6	Longitudinal case study and phenotypic multimodal characterization of McArdle disease-linked retinopathy: insight into pathomechanisms. <i>Ophthalmic Genetics</i> , 2020, 41, 73-78.	1.2	4
7	Rare Case of Transcutaneous Oxygen Desaturation in a Cancer Patient. <i>A&amp;A Practice</i> , 2019, 12, 96-98.	0.4	3
8	Diagnosis of adult-onset MELAS syndrome in a 63-year-old patient with suspected recurrent strokes – a case report. <i>BMC Neurology</i> , 2019, 19, 91.	1.8	23
9	Usefulness of Genetic Testing in Sudden Cardiac Arrest Survivors With or Without Previous Clinical Evidence of Heart Disease. <i>American Journal of Cardiology</i> , 2019, 123, 2031-2038.	1.6	30
10	Analysis of Inherited Optic Neuropathies. <i>Klinische Monatsblätter Für Augenheilkunde</i> , 2019, 236, 451-461.	0.5	4
11	A variant in <i>MRPS14</i> (uS14m) causes perinatal hypertrophic cardiomyopathy with neonatal lactic acidosis, growth retardation, dysmorphic features and neurological involvement. <i>Human Molecular Genetics</i> , 2019, 28, 639-649.	2.9	33
12	Phenotypic Spectrum of <i>HCN4</i> Mutations. <i>Circulation Genomic and Precision Medicine</i> , 2018, 11, e002033.	3.6	18
13	Unexplained cardiac arrest: a tale of conflicting interpretations of <i>KCNQ1</i> genetic test results. <i>Clinical Research in Cardiology</i> , 2018, 107, 670-678.	3.3	6
14	Movement disorders in genetically confirmed mitochondrial disease and the putative role of the cerebellum. <i>Movement Disorders</i> , 2018, 33, 146-155.	3.9	21
15	Late-onset severe long QT syndrome. <i>Annals of Noninvasive Electrocardiology</i> , 2018, 23, e12517.	1.1	1
16	Multiple clinical profiles of families with the short QT syndrome. <i>Europace</i> , 2018, 20, f113-f121.	1.7	26
17	Neutrophil extracellular trap formation requires OPA1-dependent glycolytic ATP production. <i>Nature Communications</i> , 2018, 9, 2958.	12.8	121
18	Broad phenotypes in heterozygous <i>NR5A1</i> 46,XY patients with a disorder of sex development: an oligogenic origin?. <i>European Journal of Human Genetics</i> , 2018, 26, 1329-1338.	2.8	47

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19	A novel mitochondrial ATP6 frameshift mutation causing isolated complex V deficiency, ataxia and encephalomyopathy. <i>European Journal of Medical Genetics</i> , 2017, 60, 345-351.	1.3	25
20	Characterization of two novel intronic OPA1 mutations resulting in aberrant pre-mRNA splicing. <i>BMC Medical Genetics</i> , 2017, 18, 22.	2.1	5
21	<i>SDHA</i> mutation with dominant transmission results in complex II deficiency with ocular, cardiac, and neurologic involvement. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 225-230.	1.2	26
22	Effects of endurance training on skeletal muscle mitochondrial function in Huntington disease patients. <i>Orphanet Journal of Rare Diseases</i> , 2017, 12, 184.	2.7	15
23	Mitochondrial cytopathy with common MELAS mutation presenting as multiple system atrophy mimic. <i>Neurology: Genetics</i> , 2016, 2, e121.	1.9	1
24	A novel mutation in BCS1L associated with deafness, tubulopathy, growth retardation and microcephaly. <i>European Journal of Pediatrics</i> , 2016, 175, 517-525.	2.7	14
25	The Vacuolar H <sup>+</sup> -ATPase B1 Subunit Polymorphism p.E161K Associates with Impaired Urinary Acidification in Recurrent Stone Formers. <i>Journal of the American Society of Nephrology: JASN</i> , 2016, 27, 1544-1554.	6.1	48
26	Deficiency of <scp>ECHS</scp>1 causes mitochondrial encephalopathy with cardiac involvement. <i>Annals of Clinical and Translational Neurology</i> , 2015, 2, 492-509.	3.7	90
27	Mutations in<i>SDHD</i>lead to autosomal recessive encephalomyopathy and isolated mitochondrial complex II deficiency. <i>Journal of Medical Genetics</i> , 2014, 51, 170-175.	3.2	75
28	Novel mitochondrial tRNA <sup>Ala</sup> .4282A>G gene mutation leads to chronic progressive external ophthalmoplegia plus phenotype. <i>British Journal of Ophthalmology</i> , 2014, 98, 1453-1459.	3.9	11
29	Evaluation of genome-wide loci of iron metabolism in hereditary hemochromatosis identifies PCSK7 as a host risk factor of liver cirrhosis. <i>Human Molecular Genetics</i> , 2014, 23, 3883-3890.	2.9	50
30	Mitochondrial leucine tRNA level and PTC1 are regulated in response to leucine starvation. <i>Amino Acids</i> , 2014, 46, 1775-1783.	2.7	8
31	Heterologous expression from the human D-Loop in organello. <i>Mitochondrion</i> , 2014, 17, 67-75.	3.4	7
32	Identification of SNPs in the cystic fibrosis interactome influencing pulmonary progression in cystic fibrosis. <i>European Journal of Human Genetics</i> , 2013, 21, 397-403.	2.8	20
33	Clinical and molecular characterization of the potential CF disease modifier syntaxin 1A. <i>European Journal of Human Genetics</i> , 2013, 21, 1462-1466.	2.8	8
34	Loss of insulin-induced activation of TRPM6 magnesium channels results in impaired glucose tolerance during pregnancy. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2012, 109, 11324-11329.	7.1	122
35	Sensitive and rapid detection of ganciclovir resistance by PCR based MALDI-TOF analysis. <i>Journal of Clinical Virology</i> , 2012, 54, 359-363.	3.1	23
36	qPCR-based mitochondrial DNA quantification: Influence of template DNA fragmentation on accuracy. <i>Biochemical and Biophysical Research Communications</i> , 2012, 423, 441-447.	2.1	21

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37	Impairment of mitochondrial tRNAL <sup>eu</sup> processing by a novel mutation associated with chronic progressive external ophthalmoplegia. <i>Mitochondrion</i> , 2011, 11, 488-496.	3.4	18
38	Molecular and biochemical characterisation of a novel mutation in POLG associated with Alpers syndrome. <i>BMC Neurology</i> , 2011, 11, 4.	1.8	31
39	Physiologic cold shock of <i>Moraxella catarrhalis</i> affects the expression of genes involved in the iron acquisition, serum resistance and immune evasion. <i>BMC Microbiology</i> , 2011, 11, 182.	3.3	17
40	Contiguous 16 Mb 1p36 deletion: Dominant features of classical distal 1p36 monosomy with haplo lethality. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1964-1968.	1.2	12
41	The CFTR frameshift mutation 3905insT and its effect at transcript and protein level. <i>European Journal of Human Genetics</i> , 2010, 18, 212-217.	2.8	7
42	Quantitative 1-Step DNA Methylation Analysis with Native Genomic DNA as Template. <i>Clinical Chemistry</i> , 2010, 56, 1098-1106.	3.2	24
43	Mitochondrial tRNA <sup>Leu(UUR)</sup> mutation m.3302A>G presenting as childhood onset severe myopathy: threshold determination through segregation study. <i>Journal of Inherited Metabolic Disease</i> , 2010, 33, 219-226.	3.6	10
44	Outer membrane porin M35 of <i>Moraxella catarrhalis</i> mediates susceptibility to aminopenicillins. <i>BMC Microbiology</i> , 2009, 9, 188.	3.3	13
45	Handling mammalian mitochondrial tRNAs and aminoacyl-tRNA synthetases for functional and structural characterization. <i>Methods</i> , 2008, 44, 176-189.	3.8	15
46	CLDN16 Genotype Predicts Renal Decline in Familial Hypomagnesemia with Hypercalciuria and Nephrocalcinosis. <i>Journal of the American Society of Nephrology: JASN</i> , 2008, 19, 171-181.	6.1	111
47	Mitochondrial Encephalopathy with CADASIL-Like MRI. <i>European Neurology</i> , 2007, 58, 185-188.	1.4	0
48	Rapid and reliable genotyping of polymorphic loci modifying correct splicing of CFTR pre-mRNA using mass spectrometry. <i>European Journal of Human Genetics</i> , 2007, 15, 53-61.	2.8	1
49	Mitochondrial neurogastrointestinal encephalomyopathy in three siblings. <i>Journal of Neurology</i> , 2007, 254, 146-153.	3.6	36
50	Mutations in the Tight-Junction Gene Claudin 19 (CLDN19) Are Associated with Renal Magnesium Wasting, Renal Failure, and Severe Ocular Involvement. <i>American Journal of Human Genetics</i> , 2006, 79, 949-957.	6.2	446
51	Reduced atrial connexin43 expression after pediatric heart surgery. <i>Biochemical and Biophysical Research Communications</i> , 2006, 342, 310-315.	2.1	1
52	Sodium Pump Reduction Correlates with Aortic Clamp Time in Pediatric Heart Surgery. <i>Experimental Biology and Medicine</i> , 2006, 231, 1300-1305.	2.4	2
53	Rapid typing of <i>Moraxella catarrhalis</i> subpopulations based on outer membrane proteins using mass spectrometry. <i>Proteomics</i> , 2006, 6, 172-180.	2.2	30
54	Simultaneous quantitative detection of relevant biomarkers in breast cancer by quantitative real-time PCR. <i>International Journal of Biological Markers</i> , 2006, 21, 30-39.	1.8	10

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55	Gender Modulates the Expression of Calcium-Regulating Proteins in Pediatric Atrial Myocardium. <i>Experimental Biology and Medicine</i> , 2005, 230, 853-859.	2.4	8
56	The stem-loop binding protein stimulates histone translation at an early step in the initiation pathway. <i>Rna</i> , 2005, 11, 1030-1042.	3.5	59
57	Age-dependent suppression of SERCA2a mRNA in pediatric atrial myocardium. <i>Biochemical and Biophysical Research Communications</i> , 2005, 326, 344-348.	2.1	14
58	Cardiopulmonary bypass reduces atrial Na <sup>+</sup> -ATPase expression in children. <i>Biochemical and Biophysical Research Communications</i> , 2005, 335, 700-704.	2.1	1
59	The role of common single-nucleotide polymorphisms on exon 9 and exon 12 skipping in nonmutatedCFTR Alleles. <i>Human Mutation</i> , 2004, 24, 120-129.	2.5	50
60	Functional importance of conserved nucleotides at the histone RNA 3' processing site. <i>Rna</i> , 1998, 4, 246-56.	3.5	21
61	Characterization of the Calf Thymus Hairpin-binding Factor Involved in Histone Pre-mRNA 3' End Processing. <i>Journal of Biological Chemistry</i> , 1997, 272, 10435-10441.	3.4	8
62	The gene for histone RNA hairpin binding protein is located on human chromosome 4 and encodes a novel type of RNA binding protein. <i>EMBO Journal</i> , 1997, 16, 769-778.	7.8	142